

Disease

Congenital generalized lipodystrophy type 2^{1, 2}

Gene

BSCL2

Inheritance	AR
Current Treatments	metreleptin, Tirzepatide
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	leptin level ³
Disease Frequency (per 100,000)	0.023 ⁴
Genes Included in Disease Frequency	AGPAT2, BSCL2, CAV1, CAVIN1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Generalized+Lipodystrophy&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/29704234>

² <https://pubmed.ncbi.nlm.nih.gov/39886995/>

³ <https://pubmed.ncbi.nlm.nih.gov/20097706/>

⁴ <https://www.ncbi.nlm.nih.gov/pubmed/29066925>